INTRODUCTION AND AIMS: Fanconi-Bickel Syndrome (FBS) is a rare renal disease characterized by hepato-renal glycogen accumulation and so for it is also known as Glycogenosis XI. Glycogen accumulation seems to be central in the liver and kidney dysfunction. So far, no causative treatment are available for FBS and patients can survive only by following a strict electrolytes replacement therapy. Here, we treat one patient with an innovative therapeutic approach aiming to target renal glycogen accumulation.

METHODS: One FBS patient was treated off-label for three months with a commercially available drug on the top of the electrolyte replacement therapy. Serum level of phosphate, uric acid and bicarbonate reabsorption threshold were studied as a primary endpoint. The abundance of NapiIa in urinary exosomes and glycogen content in the urinary sediment were used as innovative endpoints. All the parameters were studied at baseline, day 7, 25 and 83 of treatment and after 7 and 30 days of drug washout.

RESULTS: The treatment induced an improvement of serum phosphate and uric acid and a normalization of serum potassium level. When subjected to a bicarbonate load, the patient showed a shift to the left of the filtrate/excretion bicarbonate curve. The urine sediment showed an increase excretion of renal tubular cells, most of which were mainly from proximal tubule as showed by Napi2a staining. Glycogen content of this sediment was 3 times more abundant than in healthy subjects. The treatment induced a reduction in glycogen in the urinary sediment, while after one-month wash-out it went back to baseline level. Increasing serum phosphate level paralleled the increase Napi2a abundance in the urinary exosomes and so confirmed better tubular reabsorption.

CONCLUSIONS: We have shown here a case report in which an innovative treatment results safe and efficacious for FBS. The limited usage of a single case is the major limitation, but this can serve as background for a more extensive trial.